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Reliability of non-invasive prenatal testing questioned

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The use of non-invasive prenatal testing (NIPT) to identify chromosomal abnormalities may produce a higher rate of false positives than previously thought, according to a study published in *Genetics in Medicine*. The authors suggest that NIPT results should be interpreted with caution, and should not be considered as providing a definitive fetal diagnosis.

During pregnancy, maternal blood contains small fragments of cell-free, fetal DNA derived from the placenta. This provides a non-invasive method for testing for chromosomal abnormalities in the fetus by comparing the relative amounts of cell-free DNA from each chromosome; if the levels of DNA belonging to a specific chromosome are abnormally high, for example, it suggests the presence of an extra chromosome. Previous studies have reported conflicting results on the specificity and sensitivity of

these tests for conditions such as trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome), and trisomy 13 (Patau syndrome).

Jia-Chi Wang and colleagues analyzed 109 cases of NIPT-positive results, comparing these findings with those attained from definitive methods, such as karyotyping, a test that examines the number and structural integrity of chromosomes. They found that the rates for which results from NIPT were confirmed in the fetus ranged from 93% (38 out of 41 cases) for trisomy 21, to 38% (6 out of 16 cases) for sex chromosome abnormalities. Combining these data with the results from related studies, Wang and colleagues calculated that the positive predictive value of NIPT, which takes into account the population incidence of the specific disorders, is: 94.4% for trisomy 21, 59.5% for trisomy 18, 44.4% for trisomy 13, and 37.9% for sex chromosome abnormalities. This suggests that if a NIPT result is positive for trisomy 18, for example, the likelihood that the fetus actually has the disorder is less than 60%.

The authors note that the findings highlight the limitations of NIPT. They suggest that further studies are needed to improve the accuracy of these tests' predictive ability, and to understand the biological or technical source of the test's limitations.

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